

Claims

1. A genetic reference standard comprising at least one human genetic reference sequence
cloned into a non-mammalian animal cell line.
- 5 2. The genetic reference standard of claim 1 wherein the animal cell line is an avian cell
line.
3. The genetic reference standard of claim 2 wherein the cell line is a chicken (*Gallus*
10 *spp.*) cell line.
4. The genetic reference standard of any preceding claim wherein the cell line is a B-cell
line.
- 15 5. The genetic reference material of claim 3 wherein the chicken cell line is the chicken
DT40 cell line.
6. A genetic reference standard according to any preceding claim wherein the at least one
human genetic reference sequence is cloned into a dispensable region of the cell's
20 genome.
7. A genetic reference standard according to any preceding claim wherein the at least one
human genetic reference sequence is cloned into a non-expressed region of the cell's
genome.
- 25 8. A genetic reference standard according to any preceding claim wherein the cloned cell
line is diploid with respect to the human genetic reference sequence.
9. A genetic reference standard according to any preceding claim wherein the at least one
30 human genetic reference sequence is a plurality of human genetic reference sequences.
10. A genetic reference standard according to any preceding claim wherein the or each
human genetic reference sequence is not a functional chromosome.

11. A method of detecting a genetic variant in a sample containing human DNA comprising:

performing a test, responsive to DNA sequence, on said sample;

5 performing the same test on a reference sample embodying the genetic variant to be detected;

comparing the test results obtained from said sample and said reference sample to determine the presence or absence of said genetic variant;

10 characterised in that said reference sample is a genetic reference standard according to any preceding claim.